






















Understandings:

1. Explain what Mendel discovered about inheritance.

- Before, people always wondered what it meant by “inheritance”. They somehow noticed that the kids resembled their parents and even their grandparents. Why? Why are things like scars or cooking not inherited?

In order to find out these questions, Mendel investigated the principle of inheritance. Mendel experimented with pea plants (ugh, reminds me of my extended essay).

	Flower color	Flower position	Seed color	Seed shape	Pod shape	Pod color	Stem length
P	Purple  ×  White	Axial  ×  Terminal	Yellow  ×  Green	Round  ×  Wrinkled	Inflated  ×  Constricted	Green  ×  Yellow	Tall  ×  Dwarf
F ₁	 Purple	 Axial	 Yellow	 Round	 Inflated	 Green	 Tall

P is the parents and F₁ is the First generation, hence the offspring. The significant thing about this is that it makes sense intuitively that Purple breeding with White should give something intermediate, but did not. Sometimes, it produced offspring with different characteristics. Mendel noted down the ratio between the products.

This set the basis of genotypes, dominant, recessive and co-dominant alleles.

2. Define gametes.

- Gametes are haploid cells with 1 allele of each gene.

New offspring are produced when a male gamete and female gamete is fused. Therefore, father and mother contributes equally much genetically to the offspring. Luckily nature is not a sexist.

3. Define zygotes.

- Zygotes are the fertilized product of the fusion of male and female gamete. Now we have a diploid again.

The fertilized egg has 2 alleles per gene.

4. Explain what segregation of alleles mean.

- Segregation of alleles just means the separation of alleles.

As we saw earlier in 3.3, meiosis divides a diploid nucleus into 4 haploid nuclei. This means that if we had a gene with Tt (we have two alleles), the segregation of alleles will result in T and t in separate nuclei (and some additional variation with crossing over).

This is ultimately, as mentioned 1000 times previously, to give variations and new combinations of alleles.

5. Explain the difference between dominant, recessive, and co-dominant alleles.

- Dominant Allele: An allele that appear on the phenotype whether it is present in the homozygous or heterozygous state.

Recessive Allele: An allele that only appear on the phenotype when present in the homozygous state (two recessive).

Co-dominant Alleles: Pairs of alleles that both separately affect the phenotype (or blends to affect the phenotype when present in a heterozygote). Thus it could be dots and patterns from each parent.

However, what is the reason for this whole notion of dominant and recessive and co-dominant or incomplete dominant? Well, if the gene is a functional gene, meaning that it produces a protein that actively does things, it is dominant.

Recessive genes code for proteins those are not active. In other words, having two recessive alleles would mean that it gives off a phenotype due to the absence of a dominant allele.

6. State that many genetic diseases are due to recessive alleles of autosomal genes (we know what autosomal genes are. It is the genes that are not sex chromosomes).

- We know what a recessive allele is, and most genetic diseases are caused by recessive alleles. However, if recessive allele exists together with a dominant, it cannot function. But individuals with two recessive alleles will have an illness.

However, as long as they have the recessive allele that causes genetic disease, it can be carried on to next generation, hence we call it carrier.

	T	n
T	TT	Tn
n	Tn	nn

Here we have one unaffected, TT.

Then two carriers, Tn.

Then one with the defect, nn.

7. Outline other causes of genetic diseases, mentioning sex-linked and dominant and even co-dominant alleles.

- Some diseases are sex-linked, hence only found in combination of XX or XY. An example is red-green color blindness and hemophilia, which will be discussed later.

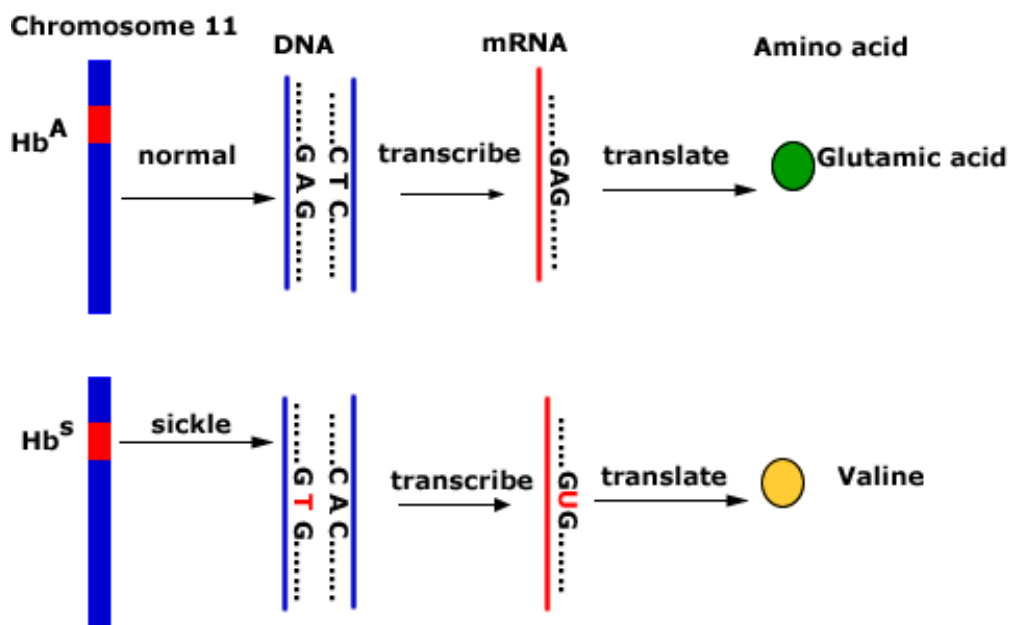
There are some cases where dominant alleles cause genetic diseases, although rare. An example of that is Huntington's disease (a neurodegenerative disorder), which will be discussed later.

Also there are diseases caused by co-dominant alleles. An example is sickle cell anemia.

Extra notes

- Sickle-cell anaemia is a disease caused by a replacement in a gene, changing the amino acid formation and hence the protein. In this particular case, it makes the RBC sickle shaped (sharp shaped). This might cause blood clotting and stroke.

On the other hand, this mutation turned out to be resistant against malaria, as the sickle shaped RBC cut through the malaria bearing cells.



It changes the amino acid glutamic acid into valine by just a change in one base. Remember this by simply the first letters. "G" comes before "V".

8. Explain what it means by sex-linkage.

- First of all, a sex-linked gene is just as it sounds. The gene is linked, or located in the sex chromosome, either X or Y. This means that there are some genes that Y chromosomes can't have but X can, and vice versa.

So how do the offspring differ between sex-linked gene and the regular genes?

Let's say we have a male with Tt and female with tt. The offspring will give the possibility of:

	T	t
t	Tt	tt
t	Tt	tt

In a regular gene, the possibility of offspring does not change if it was the female with Tt and male with tt.

But sex does matter in sex-linked genes, such as eye colour. For instance, let's say we have $X^B Y$ and $X^b X^b$. The male has brown eyes while female has blue eyes.

	X^B	Y
X^b	$X^B X^b$	$X^b Y$
X^b	$X^B X^b$	$X^b Y$

Female will have blue brown eyes only and male will have blue eyes only. If we change the situation of the parents so male has blue and female has brown, we get a different result.

	X^b	Y
X^B	$X^B X^b$	$X^B Y$
X^b	$X^b X^b$	$X^b Y$

We now have a possibility of 50% for each gender. This different inheritance pattern is called sex-linkage.

9. State some genetic diseases in humans other than the mentioned diseases.

- There are very many genetic diseases discovered, and some are phenylketonuria (PKU), Tay-Sachs disease and Marfan's syndrome.

Briefly, PKU is when an enzyme that breaks down the amino acid phenylalanine does not function. So the patient must be cautious of protein-rich food. Or else they might develop symptoms like intellectual disabilities, seizure, hyperactivity, distinct smell, etc.

Tay-Sachs is a neurodegenerative disorder. It slowly kills the neurons in the brain and the spinal cord and we can guess what will happen when those die!

Marfan's syndrome is a disease caused due to a malfunction in connective tissues (a tissue that connects different tissues and organs). This may lead to valves in heart malfunctioning hence die fast.

There are over 4000 more discovered genetic diseases. The reason why we don't see it so often is because these are recessive alleles, so probability that the same allele of two parents passing on is very small.

10. Outline some causes of mutations.

- Mutation is random, we know that. Mutations mostly happen through two causes.

The first cause is radiation that ionizes our DNA hence disrupt the bonds, and then change the sequences.

The second cause is chemical substances. The chemicals in tobacco can cause changes in DNA sequence.

However, as long as gametes are not affected, the next generation will not inherit the disease. If the gametes have been mutated, it will be passed on.

Applications and skills:

1. Know the inheritance of ABO groups.

- A, B are co-dominant, while O is recessive. We can also write it like I^A , I^B and i respectively.

Why are just A and B co-dominant while poor O is recessive? It is due to the production of glycoprotein in the membrane (for red blood cells).

I^A makes glycoprotein with acetylgalactamine.

I^B makes glycoprotein with galactose.

i makes only glycoprotein, so $I^A i$ and $I^A I^A$ will give the same A, and same with $I^B i$ and $I^B I^B$ will give B.

2. Be able to use red-green color-blindness and hemophilia as sex-linked inheritance.

- Almost all sex-linked genes are located in the X, since Y is significantly small.

Color-blindness is due to a recessive allele in X chromosome. Males have to have only 1 recessive allele to have the blindness, while females have to have two recessive alleles. Thus blindness is more common in males.

Hemophilia is also due a recessive allele in X chromosome. This causes malfunctioning of a protein involved in blood clotting. People live at max 10 years, unless the patient gets pure blood infused with functioning protein.

3. Explain the inheritance of cystic fibrosis and Huntington's disease.

- Cystic fibrosis is most common genetic disease in Europe. What it does is that a recessive allele located in chromosome 7 makes chloride channel not work properly.

So basically, patients with CF sweat out too much chloride, thus lowering the overall chloride concentration on your body. What effect does this have? Well, chlorides make an imbalance of charge, hence makes water come in by osmosis. But now that we have lack of chlorides, we won't have as much water, thus our mucus, pancreatic duct becomes thicker.

Huntington's disease is due to a dominant allele. This is still under research but we know that it degenerate the neurons in the brain.

4. Explain the consequences of Chernobyl and nuclear bombing in a biological aspect.

- Accidents that has potential to endanger DNA have serious consequences if they outbreak. The bombing and nuclear accidents have released radioactive isotopes into the unarmed society.

As these decay, they release high amount of energy that can change our DNA sequences. It may result in cancer and other deformations.

However, that is the theoretical scenario. In reality, scientists have not yet found a correlation between radiation and mutation. Though there is a possibility that mutations have undergone, it is a very small minority hence not monitored.

The Chernobyl disaster has on the other hand shown clear evidence of mutation in the surrounding. Forests have died, horses and cattle have died due to damage in thyroid gland, new ecosystem around the area has been formed, market of fishery was banned for some time, cancer rate gone up, etc.

5. Be able to draw Punnett squares and predict monohybrid genetic crosses.

- There are things that you always must include!

1. Label the gametes, such as sperm, egg, pollen, etc. Basically, label where it comes from.

2. Write the genotypes and phenotypes. An example is "Bb Brown".

3. Then state the ratio of the products (whether it is phenotypic or genotypic ratio). Sex chromosome will have ratio of 1:1 or 2:2.

Extra notes

- When you have a species and want to determine its genotype, you need to test cross. Test crossing is simply when you always cross the species with a homozygous recessive because if one breeds with a homozygous recessive it is easy to deduce the phenotypical traits.

6. Be able to compare predicted outcome versus actual outcomes of genetic crosses.

- Usually, experimental data does not fit exactly with the hypothesized data. Since genetic crosses are probability, our result might not what we expected to see.

Therefore, we have to decide whether to abandon a certain hypothesis or not by comparing predicted outcome versus actual outcome. When actual outcome is sufficiently close to predicted outcome, it can be accepted.

A technique to do that is Chi-squared test, which is in 4.1.

7. Be able to analyze Pedigree charts.

- These are just like ancestor tree, but in terms of genetics. There are a few rules to follow.

1. Squares are males, circles are females.
2. The squares/circles are colored either fully, half, or none depending on if it is an affected, carrier, or not affect individual.
3. The parents should be linked with the offspring, where it looks like a T.
4. Roman numbers = generation
5. Arabic numbers = to name individuals.

The rest is up to you how you analyze it. Keep in mind if it is the recessive or dominant that has the disease and if it is sex-linked.

TOK:

1. Mendel's theories were not accepted by the scientific community for a long time. What factors would encourage the acceptance of new ideas by the scientific community?